INTRODUCTION

CYSTINOSIS is a hereditary disorder usually appearing at 6 months of age, characterized by osteomalacia, aminoaciduria, phosphaturia, hypothyroidism and crystal deposition throughout the body tissue.

In normal humans the essential amino acid methionine is metabolized to form cysteine disulfide cross-linking of sulfahydryl groups of 2 cysteine molecules from cystine.

The cause of cystine accumulation in the lysosome is due to a defect in the lysosomal cystine transport.

In cystinotic cells the free non-protein bound cystine, being poorly soluble in an aqueous solution, accumulates in concentrations of 50 to 100 times the normal, forming crystals in many tissues and resulting in functional damage.

CASE REPORT

An 8-year old girl presented with complaints of photophobia and lacrimation of 1-month duration (Fig No.1).

On examination the visual acuity was 6/6 in each eyes, the intraocular pressure was within normal range, conjunctiva and cornea showed deposition of fine needle like crystalline deposits, involving palpebral (Fig No.2), fornical and bulbar conjunctiva and anterior 2/3rd of corneal stroma involving the peripheral more than central cornea (Fig No.3). The posterior segment examination did not show any signs of crystal deposition.

Differential diagnosis included were:

1. Cystinosis
2. Hypergammaglobulinaemia
3. Lecithin cholesterol acyl transferase deficiency
It is very unfortunate to report that patient’s parents did not make the follow up visits and change of residence led to failure of further possible contact with patient and her family.

**Discussion**

Its recessively inherited disorder classified as:

Type I or Infantile or Nephropathic Cystinosis. Type II or Juvenile or Intermediate or Adolescent Cystinosis. Type III or Adult or Benign Cystinosis.

The most important is infantile or nephropathic form, which is characterized by Fanconi syndrome with growth retardation, rickets and progressive renal failure requiring renal transplant by 10 to 13 years of age. Systemically it can involve any structure of the body like choroid plexus and brain parenchyma, eye (conjunctiva, sclera, episclera, cornea, iris, ciliary body, choroid, retinal layers especially retinal pigment epithelium, optic nerve sheath, extra ocular muscles), thyroid, liver, pancreas, spleen, bone marrow, ovaries and adrenal glands. The most common presenting complaint to ophthalmologist is photophobia due to corneal involvement and in cornea the accumulation of crystal occurs in the first year of life, initially in the periphery of the corneal stroma and with age it progress posteriorly and centripetally. By the age of 7 years the crystals are found within the or on the endothelial surface. The depth of deposition is greater in the periphery and the distribution is symmetrical between the two eyes. Histologically the crystals are birefringent, intracellular and of varying morphology.²

 Conjunctival crystals show a more whitish and amorphous appearance.

The difference in shape of crystals between different structures depends upon the tissue density in that structure, like the
appearance of corneal crystals has been attributed to tight packing of corneal lamellae. Cystinosis is frequently presented with cystine storage in the cornea and conjunctiva, and the diagnosis can be established by slit-lamp examination. It can also be confirmed by electron microscopy of a conjunctival biopsy and it also suggested that conjunctival biopsy is a valuable diagnostic tool prior to performing renal biopsy, even in cases with negative findings by ophthalmologic examination.

Studies have also shown that crystals also involve the trabecular meshwork. Ultrasonic biomicroscopy technique (UBM) shows scleral reflectivity, narrow angles, shallow anterior chamber with a reduction in the trabecular meshwork to ciliary process distance. This report of ocular UBM findings in cystinosis demonstrated narrowing of the angle and a ciliary body configuration similar to that reported for plateau iris syndrome. Gonioscopy demonstrated crystals in the trabecular meshwork. These findings may explain the predisposition of these patients to glaucoma.

Treatment of systemic involvement is symptomatic, Vitamin D therapy for rickets, thyroid hormone for hypothyroidism, fluid and electrolyte therapy for renal diseases, if fails dialysis and/or renal transplant. Cystine storage does not recur in the donor kidneys.

Ocular treatment includes cysteamine eye drops 0.5% (50mmol/L) advocated for prophylaxis, subjective improvement, and partial clearing of cornea. No ocular toxicity is documented, yet large doses of systemic administration has been shown to be cataractogenic.

Keratoplasty has been used for the symptoms but crystals may reaccumulate as early as 6 weeks or the graft remains clear. Though crystalline keratopathies are rare to encounter and diagnosis can be easily overlooked in asymptomatic cases but patient with nephropathic cystinosis especially and the possibility of crystalline ocular involvement may be considered with photophobic patients.

References


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